

REPLACEMENT SHEET

SNP Position	Reference Sequence & SNP Position Number ¹	Nucleotide Change	AA Change	Frequency in Liverpool		Number of individuals with a change in heterozygosity ²	Number of individuals with a loss in heterozygosity ³	In which Populations observed ⁴
				Blood.	Tumor			
Intron 1E	64331	A to G (TCCGTAAATTG) SEQ ID NO:3	None (+ 42 intron)	35/96 36%	36/96 38%	4	0	2, 3(N,I,A,S)
Exon 4	306382	C to G (CCCCCATACT) SEQ ID NO:4	Silent (Pro-Pro)	17/96 18%	16/94 17%	4	0	2, 3(C,I,S),6

* SNPs in Liverpool clinical tissue samples. Seen only one time and may represent sequencing artifacts. They are not included in the total counts of SNPs.

1. The SNP position number in the parenthesis is based on the beginning of each exon as 1. For SNPs within the introns, - sign was used for the ones in upstream introns and + sign for downstream introns referring the first base of the intron adjacent to the exon as 1.
2. For some heterozygosity calculations, individuals 47 and 48 were excluded because it is believed that the blood or the tumor sample was switched. These excluded cases were t=when both individuals showed a change in heterozygosity.
3. Loss of heterozygosity calculation includes any case where a heterozygous blood genotype became a homozygous genotype of the minor allele in the same individual's tumor sample. A change from a homozygous genotype of the major allele in the blood sample into a homozygous genotype of the minor allele in the tumor sample would also be counted

4. Code is as follows

- 1: SNP discovered in cDNA SNP project
- 2: SNP discovered in Liverpool DNA

3: SNP discovered in Coriell (N=Northern European, C=Chinese, I=Indo-Pakistani, A=African American, S=Southwestern Native American)

4: SNP discovered in CEPH

5: Roodi N., Bailey R., Kao W. Y., Verrier C., Yee C., Dupont W., and Parl F. J. Natl. Cancer Inst. 87 (1995) 446-451.

6: Parl, Fritz, Estrogens, Estrogen Receptor and Breast Cancer, IOS Press: Amsterdam, 2000.

Andersen TI et al. Human Mutation (1997) 9:531-536 : G to T at 838 of x03635



FIGURE 2a



REPLACEMENT SHEET

SNP Position	Reference Sequence & SNP Position #	Nucleotide Change	AA Change	Coriell Frequencies					Liverpool Frequencies	
				N.Eur	Chi	In-Pk	Af/Am	SW-NA	Blood	Tumor
Intron 1E	64331	C to T (CAATT <u>C</u> ACGGA) SEQ ID NO:5	None (+ 42 intron)	5/11 45%	0/16 0%	5/18 28%	4/18 22%	2/16 13%	35/96 36%	36/96 38%
Exon 4	306382	C to G CCCCC <u>C</u> AATACT SEQ ID NO:6	Silent (Pro-Pro)	0/8 0%	14/16 25%	14/16 25%	0/6 0%	2/13 15%	17/96 18%	16/94 17%

*, SNPs in Coriell Diversity panels. Seen only one time and may represent sequencing artifacts. They are not included in the total counts of SNPs.

FIGURE 2b



REPLACEMENT SHEET

SNP Position	Reference Sequence & SNP Position #	Nucleotide Change	AA Change	Liverpool Control Frequencies	Liverpool Frequencies	
					Blood	Tumor
Intron 1E	64331 (56346)	C to T (CAATT <u>C</u> ACGGA) SEQ ID NO:7	None (+ 51 intron)		35/96 36%	36/96 38%
Exon 4	306382 (1335)	C to G CCCCC <u>C</u> ATACT SEQ ID NO:8	Silent (Pro-Pro)		17/96 18%	16/94 17%

(blank cells mean the controls have not been genotyped for that SNP)

FIGURE 2c

REPLACEMENT SHEET

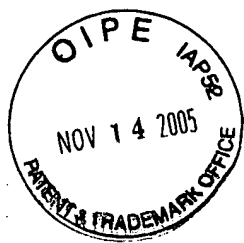


Figure 2d

PCR primers

Exon	Primer Position	PCR Product Length (bp)	Forward Primer	Reverse Primer
1E	-187/ exon1E /+163	472	AGCCAAACATTGATTCTTCAGTGCC (SEQ ID NO:9)	AAGCAACGCATGTAGAGTGCCC (SEQ ID NO:10)
4	-156/ exon4 /+103	602	GCCACTTGTGTGAACACCTTACCG (SEQ ID NO:11)	CATGTGTAATGCGTTCTTTTCCCCC (SEQ ID NO:12)